



autosomal recessive congenital stationary night blindness

Autosomal recessive congenital stationary night blindness is a disorder of the retina, which is the specialized tissue at the back of the eye that detects light and color. People with this condition typically have difficulty seeing and distinguishing objects in low light (night blindness). For example, they may not be able to identify road signs at night or see stars in the night sky. They also often have other vision problems, including loss of sharpness (reduced acuity), nearsightedness (myopia), involuntary movements of the eyes (nystagmus), and eyes that do not look in the same direction (strabismus).

The vision problems associated with this condition are congenital, which means they are present from birth. They tend to remain stable (stationary) over time.

Frequency

Autosomal recessive congenital stationary night blindness is likely a rare disease; however, its prevalence is unknown.

Genetic Changes

Mutations in several genes can cause autosomal recessive congenital stationary night blindness. Each of these genes provide instructions for making proteins that are found in the retina. These proteins are involved in sending (transmitting) visual signals from cells called rods, which are specialized for vision in low light, to cells called bipolar cells, which relay the signals to other retinal cells. This signaling is an essential step in the transmission of visual information from the eyes to the brain.

Mutations in two genes, *GRM6* and *TRPM1*, cause most cases of this condition. These genes provide instructions for making proteins that are necessary for bipolar cells to receive and relay signals. Mutations in other genes involved in the same bipolar cell signaling pathway are likely responsible for a small percentage of cases of autosomal recessive congenital stationary night blindness.

Gene mutations that cause autosomal recessive congenital stationary night blindness disrupt the transmission of visual signals between rod cells and bipolar cells or interfere with the bipolar cells' ability to pass on these signals. As a result, visual information received by rod cells cannot be effectively transmitted to the brain, leading to difficulty seeing in low light. The cause of the other vision problems associated with this condition is unclear. It has been suggested that the mechanisms that underlie night blindness can interfere with other visual systems, causing myopia, reduced visual acuity, and other impairments.

Some people with autosomal recessive congenital stationary night blindness have no identified mutation in any of the known genes. The cause of the disorder in these individuals is unknown.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- autosomal recessive complete congenital stationary night blindness
- autosomal recessive incomplete congenital stationary night blindness

Diagnosis & Management

These resources address the diagnosis or management of autosomal recessive congenital stationary night blindness:

- Genetic Testing Registry: Congenital stationary night blindness, type 1B
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1850362/>
- Genetic Testing Registry: Congenital stationary night blindness, type 1C
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750747/>
- Genetic Testing Registry: Congenital stationary night blindness, type 1D
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151193/>
- Genetic Testing Registry: Congenital stationary night blindness, type 1E
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3281215/>
- Genetic Testing Registry: Congenital stationary night blindness, type 1F
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3554399/>
- Genetic Testing Registry: Congenital stationary night blindness, type 2B
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1864877/>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Vision--Night Blindness
<https://medlineplus.gov/ency/article/003039.htm>
- Health Topic: Vision Impairment and Blindness
<https://medlineplus.gov/visionimpairmentandblindness.html>

Educational Resources

- Disease InfoSearch: Night blindness, congenital stationary, type 1b
<http://www.diseaseinfosearch.org/Night+blindness%2C+congenital+stationary%2C+type+1b/9012>
- MalaCards: autosomal recessive congenital stationary night blindness
http://www.malacards.org/card/autosomal_recessive_congenital_stationary_night_blindness
- Merck Manual Consumer Version: Structure and Function of the Eyes
<http://www.merckmanuals.com/home/eye-disorders/biology-of-the-eyes/structure-and-function-of-the-eyes>
- My46 Trait Profile
<https://www.my46.org/trait-document?trait=Congenital%20stationary%20night%20blindness&type=profile>
- Orphanet: Congenital stationary night blindness
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=215
- The University of Arizona: Congenital Stationary Night Blindness CSNB1B
<http://disorders.eyes.arizona.edu/disorders/night-blindness-congenital-stationary-csnb1b>
- The University of Arizona: Congenital Stationary Night Blindness CSNB1C
<http://disorders.eyes.arizona.edu/disorders/night-blindness-congenital-stationary-csnb1c>

Patient Support and Advocacy Resources

- American Foundation for the Blind
<http://www.afb.org/default.aspx>
- Foundation Fighting Blindness
<http://www.blindness.org/>
- Resource List from the University of Kansas Medical Center: Blind/Visual Impairment
<http://www.kumc.edu/gec/support/visual.html>
- The Foundation Fighting Blindness (Canada)
<http://ffb.ca/>

Genetic Testing Registry

- Congenital stationary night blindness, type 1B
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1850362/>
- Congenital stationary night blindness, type 1C
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750747/>
- Congenital stationary night blindness, type 1D
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151193/>
- Congenital stationary night blindness, type 1E
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3281215/>
- Congenital stationary night blindness, type 1F
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3554399/>
- Congenital stationary night blindness, type 2B
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1864877/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22autosomal+recessive+congenital+stationary+night+blindness%22+OR+%22complete+congenital+stationary+night+blindness%22+OR+%22congenital+stationary+night+blindness%22+OR+%22incomplete+congenital+stationary+night+blindness%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28congenital+stationary+night+blindness%5BTIAB%5D%29+AND+%28autosomal+recessive%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- CONE-ROD SYNAPTIC DISORDER, CONGENITAL NONPROGRESSIVE
<http://omim.org/entry/610427>
- NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1B
<http://omim.org/entry/257270>
- NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1C
<http://omim.org/entry/613216>
- NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1D
<http://omim.org/entry/613830>
- NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1E
<http://omim.org/entry/614565>
- NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1F
<http://omim.org/entry/615058>

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Reviewed: January 2014

Published: February 14, 2017

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